SGCE gene

sarcoglycan epsilon

Normal Function

The *SGCE* gene provides instructions for making a protein called epsilon (ε)-sarcoglycan, whose function is unknown. The ε-sarcoglycan protein is found within the cell membranes of the lungs, liver, kidneys, and spleen, but it is most abundant in nerve cells (neurons) in the brain and in muscle cells. Researchers suspect that in the brain the ε-sarcoglycan protein plays a role in the functioning of synapses, which are the connections between neurons where cell-to-cell communication occurs.

People inherit one copy of most genes from their mother and one copy from their father. Both copies are typically active, or "turned on," in cells. The *SGCE* gene, however, is active only when it is inherited from a person's father. This sort of parent-specific difference in gene activation is caused by a phenomenon called genomic imprinting.

Health Conditions Related to Genetic Changes

myoclonus-dystonia

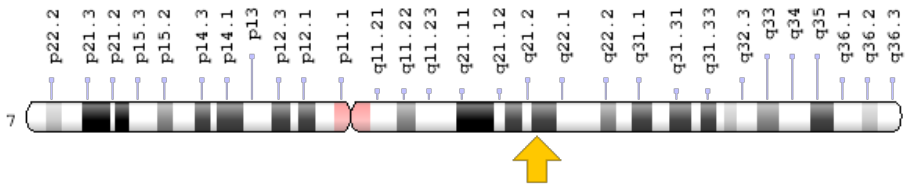
More than 65 mutations in the *SGCE* gene have been found to cause myoclonus-dystonia. Most of these mutations lead to an abnormally short, nonfunctional ε-sarcoglycan protein that is quickly broken down. Other mutations prevent the protein from reaching the cell membrane where it is needed. The protein shortage seems to affect the regions of the brain involved in coordinating movements (the cerebellum) and controlling movements (the basal ganglia). Thus, the movement problems experienced by people with myoclonus-dystonia are caused by dysfunction in the brain, not the muscles. A shortage of functional ε-sarcoglycan protein in the brain leads to the involuntary movements characteristic of myoclonus-dystonia.

Myoclonus-dystonia occurs when mutations affect the paternal copy of the *SGCE* gene. More than 95 percent of individuals who inherit an *SGCE* gene mutation from their mothers do not show symptoms of the disease. Rarely, individuals who inherit a *SGCE* gene mutation from their mothers will develop features of myoclonus-dystonia. It is unclear why a gene that is supposed to be turned off is active in these rare cases.

Chromosomal Location

Cytogenetic Location: 7q21.3, which is the long (q) arm of chromosome 7 at position 21.3

Molecular Location: base pairs 94,585,224 to 94,656,209 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DYT11
- ESG
- sarcoglycan, epsilon
- SGCE_HUMAN

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: ε-sarcoglycan https://www.ncbi.nlm.nih.gov/books/NBK6317/#A37969

GeneReviews

- Myoclonus-Dystonia https://www.ncbi.nlm.nih.gov/books/NBK1414

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SGCE%5BTIAB%5D%29+OR+%28epsilon+sarcoglycan%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- SARCOGLYCAN, EPSILON
  http://omim.org/entry/604149

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SGCE.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SGCE%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10808
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8910
- UniProt
  http://www.uniprot.org/uniprot/O43556

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